

Spinal muscular atrophy (SMA) type 1: Zolgensma[®] therapy

Zolgensma (Onasemnogene abeparvovec) is a treatment for babies or children with SMA type 1. It can also be used to treat babies with SMA before they have symptoms. This information explains Zolgensma, how it is used, and what to expect if your child has this treatment. If you have any questions or concerns, please speak to your doctor or nurse.

Spinal muscular atrophy (SMA)

SMA affects a group of nerve cells that run from the spinal cord to the muscles, called lower motor neurons. Lower motor neurons carry messages to the muscles needed to move our arms, hands, head and neck so we can roll, sit, crawl and walk. They also carry messages to the muscles we use to breathe and swallow.

SMA is caused by a fault (mutation) in a gene called survival motor neuron 1 (SMN1). We have 2 copies of this gene, one from our mother and one from our father. People with SMA have faults in both copies of their SMN1 gene.

The SMN1 gene makes the protein SMN that is needed to keep the lower motor neurons healthy. Faulty SMN1 genes cause the lower motor neurons to deteriorate so they cannot carry messages to the muscles. This makes muscles weak, and means that movement becomes difficult or impossible.

If muscles are not used they waste away (atrophy), this is why the condition is called spinal muscular atrophy.

Zolgensma

In recent years, treatments called gene therapies have been developed. These aim to correct gene faults that cause different conditions.

Zolgensma is a gene therapy that has been tested in clinical trials. It has been approved by the NHS to treat some babies and children with SMA type 1. It can also be used to treat babies who have faults in the SMN1 gene that mean they will develop SMA type 1.

Zolgensma replaces the faulty SMN1 gene with a new copy. This can stop progression of SMA type 1, but it cannot fully reverse damage to the lower motor neurons, so is not a cure.

Zolgensma uses a virus (AAV9, or adeno-associated virus 9) to deliver the new copy of the SMN1 gene into the body, and to the lower motor neurons. This virus does not cause illness and cannot make more virus copies. This means it is safe to use.

Benefits of Zolgensma

Clinical trials showed benefits of this treatment for babies and children, including:

- reaching milestones such as sitting, standing and walking, that would not have been reached without treatment
- surviving longer than expected, without needing ventilation to support their breathing
- developing speech
- feeding better, and having less need for tube feeding

A better response to treatment is likely to be seen if a baby can be treated before they have severe muscle weakness.

Having Zolgensma

Zolgensma is a liquid. It is given through a thin, plastic tube (cannula) that is put into a vein, this is called an infusion. It is given over about 1 hour. The medical team usually put 2 cannulas in, so if 1 does not work or falls out during the infusion it can be swapped over straight away. Your child will be monitored closely by a nurse during the infusion.

Before treatment

Your child will need some blood tests before treatment. This is so the doctors can check it is safe to treat your child with Zolgensma. These blood tests will:

- check that your child does not have antibodies to the AAV9 virus, which would mean the treatment would not work
- check that your child's kidneys, liver, heart and blood clotting are normal

The doctor might want your child to have more tests before starting treatment. They will talk to you about any tests your child will have, and the results. All children will need to have a physiotherapy assessment before having the treatment.

A week before treatment

You will come to the hospital for the day about 1 week before treatment, so your child can be seen by a nurse and doctor. They will usually have a blood test and heart scan at this visit. You will usually be asked to sign a consent form for the Zolgensma treatment at this visit.

The day before treatment

Your child will be admitted to the day case unit, where they will be seen by a nurse and a doctor. They will be given a steroid medicine called prednisolone, which lowers the chance of any treatment side effects. The prednisolone will be given by mouth. It can also be given by a tube if your child has a nasogastric or gastrostomy feeding tube.

Your child will stay in hospital the night before their treatment.

After treatment

For the first 48 hours after treatment, your child will stay in hospital or in accommodation close by. This is to make sure your child stays well after treatment. After your child is sent home from hospital (discharged), they will continue to be followed up in their usual neuromuscular centre.

Your child will need to take prednisolone for at least 30 days after treatment. Your doctor will give you a plan to continue this medicine, or to gradually reduce the amount (dose) your child takes over time. It is really important that you **do not stop this medicine** without speaking to a doctor or nurse caring for your child.

When to contact the medical team

After treatment, you must contact your child's medical team straight away if they:

- have any yellowing of the skin, or the whites of their eyes (jaundice)
- are being sick (vomiting) for no reason
- seem to have tummy pain
- have blood in their poo, or vomit blood
- have unexpected bruising or bleeding
- have any symptoms of TMA (fits, fewer wet nappies or peeing less than usual)
- are in contact with anyone with chicken pox or measles
- seem to have difficulties breathing or swallowing

Side effects of prednisolone (steroid medicine)

Steroids have some side effects that will need to be monitored, such as:

- increased appetite
- weight gain
- mood changes
- raised blood pressure
- an increase in blood sugar levels

If your child has tummy pain, any sign of blood in their poo, or they vomit blood you should contact your medical team.

Steroids can also make you more likely to get an infection. Your child should not be in contact with anyone with chicken pox or measles while they have this medicine. If this happens, speak to your medical team straight away. These infections can be serious in people taking prednisolone, but there are treatments.

Steroids can also cause stomach irritation. Do not give your child ibuprofen (Nurofen) regularly whilst they are taking prednisolone. The doctors and nurses will make sure your GP and local knows that prednisolone has been prescribed.

You might be given a steroid emergency card to carry with you. Take this card to other appointments or the emergency department (A&E) to make sure health professionals know your child is taking steroids.

Side effects of Zolgensma

There can be side effects of Zolgensma. These are most likely to happen in the first 6 weeks after treatment. Your child will have blood tests to check for any side effects. These will be at least once every week to begin with, and then every 2 weeks until your doctor and nurse are confident that there are no side effects. You will be given details about these blood tests before you are sent home from hospital.

Zolgensma is a new treatment. This means that we do not know how long it will work to control the symptoms of SMA. It is possible that there could be rare side effects that have not been found yet, or that it could cause side effects after many years (long-term side effects). Your doctor will talk to you about the possible side effects, and what to look out for.

Feeling and being sick (vomiting) is a common side effect. Your doctor can give your child anti-sickness medicine to help.

Flu-like illness. Your child might develop a high temperature (fever) after the infusion. This can be managed with paracetamol. In the first few days after treatment many children are tired, and might sleep more than usual.

Effects on the liver. The prednisolone medicine helps to stop any liver damage. Your child will have regular blood tests to check how their liver is working.

Bruising and bleeding. Platelets in the blood help us to form scabs and blood clots. If the number of platelets in the blood is too low, this can cause bruising and bleeding. Zolgensma can sometimes cause a drop in the number of platelets. This will be monitored by blood tests.

Thrombotic microangiopathy (TMA) is a rare condition related to a low platelet count. It has been reported about 1 week after Zolgensma infusion. In March 2021, there had only been 5 cases reported out of about 800 children treated.

Symptoms include bruising, bleeding, feeling sleepy and sometimes fits (seizures). You might notice fewer wet nappies, or that your child is peeing less. If your child has any of these symptoms get urgent medical advice from your medical team. Go to your nearest emergency department (A&E) if you cannot contact your medical team (for example on the weekend or overnight).

More serious side effects

Very occasionally, people have a more serious reaction after Zolgensma. This causes inflammation of multiple organs, such as the kidneys, liver and heart. If this happens, treatment in the intensive care unit is needed. Your child will have regular blood tests to check their kidneys, liver and heart.

If you are worried that your child is not well, you should contact the medical team (see page 5).

Other things to know about

Vaccinations

Some vaccines use 'live' viruses. These **should not** be given whilst your child is taking prednisolone, or for the first 3 months after they stop prednisolone. Your doctor or nurse will review your child's vaccination plans and tell you when these can safely be given.

Zolgensma in pee and poo

Small amounts of Zolgensma, which includes the AAV9 virus, can be found in the urine (pee) and poo. Good hand hygiene is important for 1 month after treatment. Wash your hands thoroughly after changing nappies. You could also wear gloves. Nappies must be double sealed in bags and then thrown out in the regular rubbish bin.

Pregnancy and Zolgensma

It is recommended that pregnant women avoid coming into contact with Zolgensma. Your child should have less contact with pregnant women when they are having this treatment, if this is possible. If you have any questions or concerns about this, speak to your doctor or nurse.

Spinraza®

If your child has previously had a treatment called Spinraza (nusinersen) or risdiplam this will be stopped if they have Zolgensma.

Other treatment options (alternatives)

There is a treatment called Spinraza, which works in a different way to Zolgensma. This is given by repeated injections into the lower part of the back. Your doctor will help you to decide which treatment is best for your child.

It is important to know that whether or not your child has Zolgensma treatment, there are ways to manage symptoms, reduce complications of muscle weakness and maintain the best quality of life for your child.

Physiotherapy assessment will be a very important part of your child's care. They will also see other health professionals, such as speech and language therapists, dietitians and occupational therapists.

Follow-up appointment

For the first few months after treatment, your child will need regular blood tests and assessments. You will be given details about these appointments before being leaving hospital.

These appointments will be at Evelina London Children's Hospital, with a doctor and physiotherapist. Your child will usually have an appointment once a month for 3 months, at 4 weeks, 8 weeks and 12 weeks after treatment.

After the first few months, they will usually have an appointment at least 2 to 3 times a year. These appointments might be at your usual neuromuscular centre.

Useful sources of information

SMAUK (Spinal muscular atrophy UK) has information and support for families affected by SMA, [phone](tel:01789267520) 01789267520 [web](http://www.smauk.org.uk) www.smauk.org.uk

The NHS website has information on SMA.

[web](http://www.nhs.uk/conditions/spinal-muscular-atrophy-sma) www.nhs.uk/conditions/spinal-muscular-atrophy-sma

Contact us

If you have any questions or concerns about Zolgensma, contact the **neuromuscular team**, Monday to Friday, 9am to 5pm.

- Clinical nurse specialists (CNS), [phone](tel:02071885944) 020 7188 5944
- Consultants, [phone](tel:02071883998) 020 7188 3998 or 020 7188 4006 or 0207 7188 4648

In emergencies, please contact 111 or go to your nearest emergency department (A&E).

For the **24-hour paediatric neurology service**, contact St Thomas' Hospital.

[phone](tel:020771887188) 0207 7188 7188 and ask for the bleep desk, then the paediatric neurology registrar.

For more information on conditions, procedures, treatments and services offered at our hospitals, please visit [web](http://www.evelinalondon.nhs.uk/leaflets) www.evelinalondon.nhs.uk/leaflets

Evelina London Medicines Helpline

If you have any questions or concerns about your child's medicines, please speak to the staff caring for them or contact our helpline, [phone](tel:02071883003) 020 7188 3003 Monday to Friday, 10am to 5pm, [email](mailto:letstalkmedicines@gstt.nhs.uk) letstalkmedicines@gstt.nhs.uk



Patient Information Forum

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